

ABSTRACT OF THE INVENTION

The invention provides methods and compositions for diagnosing and treating ectodermal disorders, particularly ectodermal dysplasia syndromes such as Clouston syndrome, which are associated with misexpression of a TAJ gene. Methods detect the presence of or predisposition to an ectodermal disorder by (a) detecting the presence of a TAJ gene or gene product in a cell; and (b) correlating the presence of the TAJ gene or gene product with a presence of or predisposition to an ectodermal disorder. Other methods modulate the functional expression of a TAJ gene or gene product in a cell comprising the step(s) of contacting a cell with an agent which specifically binds and modulates the functional expression of a TAJ gene or gene product, wherein (a) the cell is an ectodermal cell; or (b) the cell is a germ cell which gives rise to progeny ectodermal cells and detecting the functional expression of the TAJ gene or gene product in the progeny cells.